FUCA1 gene

fucosidase, alpha-L- 1, tissue

Normal Function

The *FUCA1* gene provides instructions for making an enzyme called alpha-L-fucosidase. This enzyme is found in lysosomes, which are compartments in the cell that digest and recycle materials. Within lysosomes, this enzyme plays a role in the breakdown of complexes of sugar molecules (oligosaccharides) attached to certain proteins (glycoproteins) and fats (glycolipids). Alpha-L-fucosidase is responsible for cutting (cleaving) off a sugar molecule called fucose toward the end of the breakdown process.

Health Conditions Related to Genetic Changes

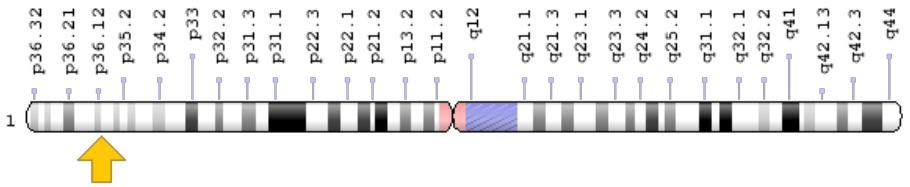
fucosidosis

At least 26 mutations in the *FUCA1* gene have been found to cause fucosidosis. Most of these mutations result in an abnormally short, nonfunctional alpha-L-fucosidase enzyme. Without this enzyme, glycolipids and glycoproteins cannot be completely broken down. These partially broken down compounds accumulate in the lysosomes and cause cells throughout the body to malfunction. Brain cells are particularly sensitive to the buildup of glycolipids and glycoproteins, which can result in cell death. Loss of brain cells is thought to cause the neurological symptoms of fucosidosis. Accumulation of glycolipids and glycoproteins also occurs in other organs such as the liver, spleen, skin, heart, pancreas, and kidneys, contributing to the additional symptoms of fucosidosis.

Chromosomal Location

Cytogenetic Location: 1p36.11, which is the short (p) arm of chromosome 1 at position 36.11

Molecular Location: base pairs 23,845,077 to 23,868,369 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- alpha-L-fucosidase
- alpha-L-fucosidase 1
- FUCO_HUMAN

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Glycoprotein disorders result from defects in lysosomal hydrolases
  https://www.ncbi.nlm.nih.gov/books/NBK28215/#A2910

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28FUCA1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- FUCOSIDASE, ALPHA-L, 1
  http://omim.org/entry/612280

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_FUCA1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=FUCA1%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4006
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2517
- UniProt
  http://www.uniprot.org/uniprot/P04066

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